TCHH gene

trichohyalin

Normal Function

The *TCHH* gene provides instructions for making a protein called trichohyalin. This protein is primarily found in hair follicles, which are specialized structures in the skin where hair growth occurs. Trichohyalin can also be found in the hair strand (shaft). Once trichohyalin is produced, it is modified by other proteins so that it can attach (bind) to other trichohyalin proteins and to molecules called keratin intermediate filaments to create organized cross-links. These links form dense networks that give the hair shaft its cylindrical shape.

Health Conditions Related to Genetic Changes

uncombable hair syndrome

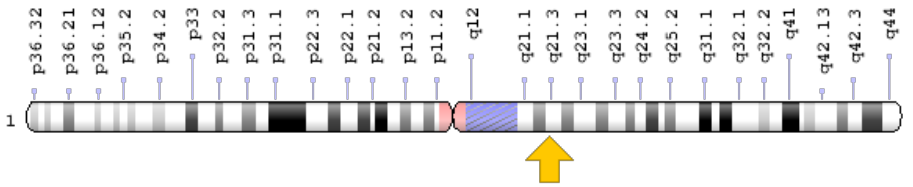
At least one mutation in the *TCHH* gene has been found to cause uncombable hair syndrome. This condition is characterized by dry, frizzy, blond scalp hair that cannot be combed flat. This condition usually improves over time, and by adolescence individuals with uncombable hair syndrome have hair that lies flat and has normal or nearly normal texture.

The *TCHH* gene mutation that has been identified leads to a premature stop signal in the instructions used to make trichohyalin, resulting in an abnormally short protein with severely reduced activity. A shortage (deficiency) of functional protein reduces the cross-links that are formed between trichohyalin proteins and keratin intermediate filaments. As a result, the cross-section of the hair shaft becomes triangular, heart-shaped, or flat. These angular hair shafts result in frizzy hair that will not lie flat, which is typical of uncombable hair syndrome.

Chromosomal Location

Cytogenetic Location: 1q21.3, which is the long (q) arm of chromosome 1 at position 21.3

Molecular Location: base pairs 152,106,317 to 152,115,454 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AHF
- THH
- THL
- TRHY

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Cutaneous Appendages https://www.ncbi.nlm.nih.gov/books/NBK10037/#A2933
- Madame Curie Bioscience: Molecular Mechanisms of Embryonic and Adult Hair Development https://www.ncbi.nlm.nih.gov/books/NBK6571/#A62117

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TCHH%5BTIAB%5D%29+OR+%28trichohyalin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- TRICHOHYALIN
  http://omim.org/entry/190370

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TCHH%5Bgene%5D
- HGNC Gene Family: EF-hand domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/863
- HGNC Gene Family: S100 fused type protein family
  http://www.genenames.org/cgi-bin/genefamilies/set/1350
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11791
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7062
- UniProt
  http://www.uniprot.org/uniprot/Q07283

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